

Human Genetic Variation

About the National Institutes of Health

The National Institutes of Health (NIH), the world's top medical research center, is charged with addressing the health concerns of the nation. The NIH is the largest U.S. governmental sponsor of health studies conducted nationwide.

Simply described, the NIH's goal is to acquire new knowledge to help prevent, detect, diagnose, and treat disease and disability, from the rarest genetic disorder to the common cold. The NIH works toward that goal by conducting research in its own laboratories in Bethesda, Maryland, and at several other locations throughout the United States; supporting the research of nonfederal scientists throughout the country and abroad; helping to train research investigators; and fostering communication of medical information to the public.

The NIH Supports Research

A principal concern of the NIH is to invest wisely the tax dollars entrusted to it for the support and conduct of medical research. Approximately 82 percent of the investment is made through grants and contracts supporting research and training in more than 2,000 universities, medical schools, hospitals, and research institutions throughout the United States and abroad.

Approximately 10 percent of the budget goes to more than 2,000 projects conducted mainly in NIH laboratories. About 80 percent covers support costs of research conducted both within and outside the NIH.

NIH Research Grants

To apply for a research grant, an individual scientist must submit an idea in a written application. Each application undergoes a peer review process. A panel of scientific experts, who are active researchers in the medical sciences, first evaluates the scientific merit of the application. Then, a national advisory council or board, composed of eminent scientists as well as public members who are interested in health issues or the medical sciences, determines the project's overall merit and priority. Because funds are limited, the process is very competitive.

The Nobelists

The rosters of those who have conducted research, or who have received NIH support over the years, include some of the world's most illustrious scientists and physicians. Among them are 97 scientists who have won Nobel Prizes

About the National Human Genome Research Institute

The National Human Genome Research Institute (NHGRI) is leading the international effort to identify and characterize the estimated 80,000 to 100,000 genes that orchestrate a single cell's development into a human infant and then an adult, and that govern whether that individual will be susceptible to diseases such as muscular dystrophy, cancer, Alzheimer disease, high blood pressure, and obesity.

Part of the National Institutes of Health, the federal government's biomedical research arm, NHGRI has set the year 2003 as its deadline for completing the DNA sequence of the human genome, our genetic blueprint.

Completing the sequence of the human genome and deciphering its functions are the first step toward "molecular medicine," the revolutionary approach to diagnosis and treatment that will create targeted, individualized health care in the early 21st century. Then, each person should be able to determine his or her risk for disease through genetic tests. If the tests indicate increased susceptibility to a disease, the individual will be able to obtain counseling on how to reduce that risk—perhaps by periodic medical check-ups, a special diet and other lifestyle changes, and drugs tailored to his or her genetic profile. Treatment of disease also likely will include gene therapies to replace, compensate for, or repair the genes that play a role in the disease.

In addition to genetics research, NHGRI sponsors research exploring the potential ethical, legal, and social consequences of the anticipated genetics revolution in medicine. By focusing now on preventing the potential misuses of genetic information in insurance and employment, NHGRI is helping ensure that genetic information will be used as it was intended: to promote human health and save lives.

for achievements as diverse as deciphering the genetic code and learning what causes hepatitis.

Five Nobelists made their prize-winning discoveries in NIH laboratories: Doctors Christian B. Anfinsen, Julius Axelrod, D. Carleton Gajdusek, Marshall W. Nirenberg, and Martin Rodbell.

For more information about the National Human Genome Research Institute, visit its Web site at: <http://www.nhgri.nih.gov>.

Impact of the NIH on the Nation's Health

The research programs of the NIH have been remarkably successful during the past 50 years. NIH-funded scientists have made substantial progress in understanding the basic mechanisms of disease and have vastly improved the preventive, diagnostic, and therapeutic options available.

During the last few decades, NIH research played a major role in making possible achievements like these:

- Mortality from heart disease, the number one killer in the United States, dropped by 36 percent between 1977 and 1999.
- Improved treatments and detection methods increased the relative five-year survival rate for people with cancer to 60 percent.
- Those suffering from depression now look forward to returning to work and leisure activities, thanks to treatments that give them an 80 percent chance to resume a full life in a matter of weeks.
- Vaccines protect against infectious diseases that once killed and disabled millions of children and adults.
- In 1990, NIH researchers performed the first trial of gene therapy in humans. Scientists are increasingly able to locate, identify, and describe the functions of many of the genes in the human genome. The ultimate goal is to develop screening tools and gene therapies for the general population for cancer and many other diseases.

Educational and Training Opportunities at the NIH

The NIH offers myriad opportunities including summer research positions for students. For details, visit <http://science.education.nih.gov/students>.

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The NIH Office of Science Education

The NIH Office of Science Education (OSE) is bringing exciting new resources free of charge to science teachers of grades kindergarten through 12. OSE learning tools support teachers in training the next generation of scientists and scientifically literate citizens. These materials cover information not available in standard textbooks and allow students to explore biological concepts using real world examples. In addition to the curriculum supplements, OSE provides a host of valuable resources accessible through the OSE Web site <http://science.education.nih.gov/>, such as:

- **Snapshots of Science and Medicine.**² This online magazine—plus interactive learning tools—is designed for ease of use in high school science classrooms. Three issues, available for free, are published during the school year. Each focuses on a new area of research and includes four professionally written articles on findings, historical background, related ethical questions, and profiles of people working in the field. Also included are a teaching guide, classroom activities, handouts, and more. (<http://science.education.nih.gov/snapshots>)
- **Women Are Scientists Video and Poster Series.**³ This series provides teachers and guidance counselors with free tools to encourage young women to pursue careers in the medical field. The informative, full-color video and poster sets focus on some of the careers in which women are currently underrepresented. The first set, titled “Women are Surgeons,” has been completed. The second, “Women are Pathologists,” will be finished in 2000, and the third, “Women are Researchers,” in 2001. (<http://science.education.nih.gov/women>)
- **Internship Programs.** Visit the OSE Web site to obtain information on a variety of NIH programs open to

teachers and students. (<http://science.education.nih.gov/students>)

- **National Science Teacher Conferences.** Thousands of copies of NIH materials are distributed to teachers for free at the OSE exhibit booth at conferences of the National Science Teachers Association and the National Association of Biology Teachers. OSE also offers teacher-training workshops at many conferences. (<http://science.education.nih.gov/exhibits>)

In the development of learning tools, OSE supports science education reform as outlined in the *National Science Education Standards* and related guidelines.

We welcome your comments about existing resources and suggestions about how we may best meet your needs. Feel free to send your comments to us at <http://science.education.nih.gov/feedback>.

^{2,3} These projects are collaborative efforts between OSE and the NIH Office of Research on Women's Health.